

SHORT GCG EXPANSIONS IN THE PAB II GENE FOR OCULO-
PHARYNGEAL MUSCULAR DYSTROPHY AND DIAGNOSTIC THEREOF

ABSTRACT OF THE DISCLOSURE

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The present invention relates to a human PAB II gene containing transcribed
5 polymorphic GCG repeat, which comprises a sequence as set forth in SEQ ID NO:3,
which includes introns and flanking genomic sequence. The allelic variants of GCG
repeat of the human PAB II gene are associated with a disease related with protein
accumulation in nucleus, such as polyalanine accumulation, a disease related with
swallowing difficulties, such as oculopharyngeal muscular dystrophy. The present
10 invention also relates to a method for the diagnosis of a disease with protein
accumulation in nucleus, which comprises the steps of: a) obtaining a nucleic acid
sample of said patient; and b) determining allelic variants of GCG repeat of the gene of
claim 1, and wherein long allelic variants are indicative of a disease related with
protein accumulation in nucleus.

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